

# Health Care Provider Fact Sheet

## Disease Name

## Carnitine uptake deficiency

### Alternate name(s)

Systemic carnitine deficiency, Carnitine deficiency, Carnitine transporter deficiency

### Acronym

SCD, CUD

### Disease Classification

Fatty Acid Oxidation Disorder

### Variants

N/A

### Variant name

N/A

### Symptom onset

Infancy or childhood with fasting hypoglycemia, weakness and/or cardiomyopathy.

### Symptoms

Hypoketotic hypoglycemia, seizures, vomiting, lethargy progressing to coma. Chronic muscle weakness, cardiomyopathy, hepatomegaly.

### Natural history without treatment

Non-progressive developmental delay due to hypoglycemia, cardiomyopathy and muscle weakness.

### Natural history with treatment

Developmental delay, if present, is not reversed by treatment. Cardiomyopathy and muscle weakness can be reversed by treatment.

### Treatment

Carnitine supplementation, no fasting.

### Emergency Medical Treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website:

<http://www.acmg.net/StaticContent/ACT/C0.pdf>

### Physical phenotype

Cardiomyopathy, muscle weakness.

### Inheritance

Autosomal recessive

### General population incidence

1/40,000

### Ethnic differences

No

### Population

N/A

### Ethnic incidence

N/A

### Enzyme location

Muscle, heart, kidney, leukocytes and fibroblasts

### Enzyme Function

Transports carnitine into cells

### Missing Enzyme

Carnitine transporter

### Metabolite changes

Decreased free carnitine in plasma, increased carnitine in urine, decreased carnitine in muscle.

### Prenatal testing

Protein analysis in cultured amniocytes, biochemical analyte testing. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS or amniocytes is possible.

### MS/MS Profile

Reduced concentrations of free carnitine and various acylcarnitine species.

### OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=212140>

### Genetests Link

[www.genetests.org](http://www.genetests.org)

### Support Group

FOD Family Support Group

<http://www.fodsupport.org>

Save Babies through Screening Foundation

<http://www.savebabies.org>

Genetic Alliance

<http://www.geneticalliance.org>

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